



COQ8A gene

coenzyme Q8A

Normal Function

The *COQ8A* gene provides instructions for making a protein that is involved in the production of a molecule called coenzyme Q10, which has several critical functions in cells throughout the body. In cell structures called mitochondria, coenzyme Q10 plays an essential role in a process called oxidative phosphorylation, which converts the energy from food into a form cells can use. Coenzyme Q10 is also involved in producing pyrimidines, which are building blocks of DNA, its chemical cousin RNA, and molecules such as ATP and GTP that serve as energy sources in the cell. In cell membranes, coenzyme Q10 acts as an antioxidant, protecting cells from damage caused by unstable oxygen-containing molecules (free radicals), which are byproducts of energy production.

Health Conditions Related to Genetic Changes

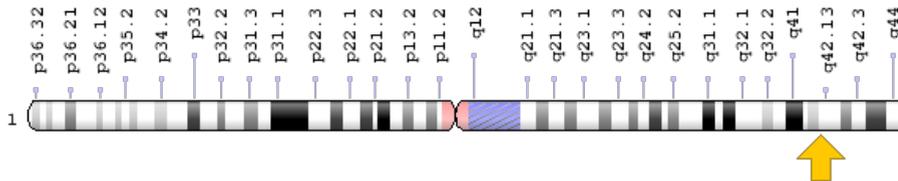
primary coenzyme Q10 deficiency

At least 36 mutations in the *COQ8A* gene have been found to cause a disorder known as primary coenzyme Q10 deficiency. This rare disease usually becomes apparent in infancy or early childhood, but it can occur at any age. It can affect many parts of the body, most often the brain, muscles, and kidneys. The *COQ8A* gene mutations associated with this disorder change the structure of the *COQ8A* protein or prevent its production, which impairs the normal production of coenzyme Q10. Studies suggest that a shortage (deficiency) of coenzyme Q10 impairs oxidative phosphorylation and increases the vulnerability of cells to damage from free radicals. A deficiency of coenzyme Q10 may also disrupt the production of pyrimidines. These changes can cause cells throughout the body to malfunction, which may help explain the variety of organs and tissues that can be affected by primary coenzyme Q10 deficiency.

Chromosomal Location

Cytogenetic Location: 1q42.13, which is the long (q) arm of chromosome 1 at position 42.13

Molecular Location: base pairs 226,939,339 to 226,987,545 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- aarF domain-containing protein kinase 3
- ADCK3
- ARCA2
- atypical kinase COQ8A, mitochondrial
- CABC1
- chaperone activity of bc1 complex-like, mitochondrial
- chaperone, ABC1 activity of bc1 complex homolog
- coenzyme Q protein 8A
- coenzyme Q8 homolog
- COQ8
- COQ10D4
- SCAR9

Additional Information & Resources

Educational Resources

- Linus Pauling Institute: Coenzyme Q10
<http://lpi.oregonstate.edu/mic/dietary-factors/coenzyme-Q10>
- Molecular Biology of the Cell (fourth edition, 2002): How Cells Obtain Energy from Food
<https://www.ncbi.nlm.nih.gov/books/NBK26882/>
- The Cell: A Molecular Approach (second edition, 2000): The Mechanism of Oxidative Phosphorylation
<https://www.ncbi.nlm.nih.gov/books/NBK9885/>

GeneReviews

- Primary Coenzyme Q10 Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK410087>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28COQ8A%5BTIAB%5D%29+OR+%28coenzyme+Q8A%5BTIAB%5D%29%29+OR+%28%28ADCK3%5BTIAB%5D%29+OR+%28ARCA2%5BTIAB%5D%29+OR+%28CABC1%5BTIAB%5D%29+OR+%28COQ10D4%5BTIAB%5D%29+OR+%28COQ8%5BTIAB%5D%29+OR+%28SCAR9%5BTIAB%5D%29+OR+%28aarF+domain-containing+protein+kinase+3%5BTIAB%5D%29+OR+%28atypical+kinase+COQ8A,+mitochondrial%5BTIAB%5D%29+OR+%28chaperone+activity+of+bc1+complex-like,+mitochondrial%5BTIAB%5D%29+OR+%28chaperone,+ABC1+activity+of+bc1+complex+homolog%5BTIAB%5D%29+OR+%28coenzyme+Q+protein+8A%5BTIAB%5D%29+OR+%28coenzyme+Q8+homolog%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- COENZYME Q8A
<http://omim.org/entry/606980>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ADCK3.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=COQ8A%5Bgene%5D>

- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16812
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/56997>
- UniProt
<http://www.uniprot.org/uniprot/Q8NI60>

Sources for This Summary

- Acosta MJ, Vazquez Fonseca L, Desbats MA, Cerqua C, Zordan R, Trevisson E, Salviati L. Coenzyme Q biosynthesis in health and disease. *Biochim Biophys Acta*. 2016 Aug;1857(8):1079-85. doi: 10.1016/j.bbabbio.2016.03.036. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/27060254>
- OMIM: COENZYME Q8A
<http://omim.org/entry/606980>
- Cullen JK, Abdul Murad N, Yeo A, McKenzie M, Ward M, Chong KL, Schieber NL, Parton RG, Lim YC, Wolvetang E, Maghzal GJ, Stocker R, Lavin MF. AarF Domain Containing Kinase 3 (ADCK3) Mutant Cells Display Signs of Oxidative Stress, Defects in Mitochondrial Homeostasis and Lysosomal Accumulation. *PLoS One*. 2016 Feb 11;11(2):e0148213. doi: 10.1371/journal.pone.0148213. Erratum in: *PLoS One*. 2016;11(7):e0160162.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/26866375>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4751082/>
- Desbats MA, Lunardi G, Doimo M, Trevisson E, Salviati L. Genetic bases and clinical manifestations of coenzyme Q10 (CoQ 10) deficiency. *J Inherit Metab Dis*. 2015 Jan;38(1):145-56. doi: 10.1007/s10545-014-9749-9. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25091424>
- Doimo M, Desbats MA, Cerqua C, Cassina M, Trevisson E, Salviati L. Genetics of coenzyme q10 deficiency. *Mol Syndromol*. 2014 Jul;5(3-4):156-62. doi: 10.1159/000362826.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25126048>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4112527/>
- GeneReview: Primary Coenzyme Q10 Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK410087>
- Khadria AS, Mueller BK, Stefely JA, Tan CH, Pagliarini DJ, Senes A. A Gly-zipper motif mediates homodimerization of the transmembrane domain of the mitochondrial kinase ADCK3. *J Am Chem Soc*. 2014 Oct 8;136(40):14068-77. doi: 10.1021/ja505017f.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25216398>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4195374/>

- Mignot C, Apartis E, Durr A, Marques Lourenço C, Charles P, Devos D, Moreau C, de Lonlay P, Drouot N, Burglen L, Kempf N, Nourisson E, Chantot-Bastaraud S, Lebre AS, Rio M, Chaix Y, Bieth E, Roze E, Bonnet I, Canaple S, Rastel C, Brice A, Rötig A, Desguerre I, Tranchant C, Koenig M, Anheim M. Phenotypic variability in ARCA2 and identification of a core ataxic phenotype with slow progression. *Orphanet J Rare Dis.* 2013 Oct 28;8:173. doi: 10.1186/1750-1172-8-173.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24164873>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3843540/>
 - Stefely JA, Reidenbach AG, Ulbrich A, Oruganty K, Floyd BJ, Jochem A, Saunders JM, Johnson IE, Minogue CE, Wrobel RL, Barber GE, Lee D, Li S, Kannan N, Coon JJ, Bingman CA, Pagliarini DJ. Mitochondrial ADCK3 employs an atypical protein kinase-like fold to enable coenzyme Q biosynthesis. *Mol Cell.* 2015 Jan 8;57(1):83-94. doi: 10.1016/j.molcel.2014.11.002.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/25498144>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4289473/>
-

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/COQ8A>

Reviewed: April 2017
Published: April 25, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services